PWG Genetics and Society workshop (during ECA meeting)
June 29, 2013
Dublin (Ireland)

The PWG Genetics and Society held a workshop on Saturday 29th June on 'reporting cytogenetic results'. There was an overview of what basic requirements are needed in a cytogenetic report according to the European Cytogenetic guidelines, the OECD guidelines (2007) and ISO15189.

- Patient demographics, sample type + origin;
- Sample receipt and report dates;
- Referral reason i.e. clinical reason for requesting test;
- Probe/kit manufacturer;
- Correct use of ISCN 2013;
- Written description of the abnormalities.

INTERPRETATION

- Correlation of result to referral reason/question;
- Expected clinical outcome;
- Correct etiology;
- Request for appropriatee follow up samples where applicable;
    - Parental bloods?
    - Repeat PND sample?
    - Additional studies required e.g. microarrays?
- Assessment of recurrence/PND in future pregnancies;
- Prognostic indicators/limitations of test;
- Referral for Genetic counselling.

Examples of reports from recent external quality assessment schemes (CEQA and UK NEQAS for clinical cytogenetics), highlighted that there was still a proportion of laboratories (10-20% depending on EQA) who omitted essential information in their reports. While the omission detailed below will not be applicable to every diagnostic case, professional judgement should be used on a case by case basis, taking into account the referral reason and the abnormality detected.

Interpretative information missing on reports included:

- Relating the result to the referral reason/clinical question;
- Expected clinical outcome;
- Whether there is a risk of recurrence in the future;
- Recommendation for prenatal diagnosis;
- Referral to a clinical geneticist.

Technical information sometimes missing on the array reports included:

- Genome build;
- Array platform;
- Limitations of the test.

This talk was followed by a 'hands-on' workshop where the participants were split into 4 groups and given 4 different case reports to assess. The four cases included an interstitial deletion (G-banding and FISH); a derivative chromosome from an inherited translocation (G-banding and FISH); a recurrent translocation in a lymphoma and a chromosome loss detected on arrays. At the end of the session the participants from each group fed back on the missing elements for all 4 cases and there was an overview of all the omissions.

The workshop was well received by all the participants.

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